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NEUROFIBROMATOSIS ASSOCIATED WITH HYPERVASCULARITY- A CASE REPORT

Dermatology

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ABSTRACT

Neurofibromatosis uncommonly presents with hypervascular neurofibromas, probably due to elevated angiogenic factors. We report a case of neurofibromatosis with a proliferative vascular growth at the site of previously biopsied neurofibroma and revealed to have increased vasculature on histopathological examination.

KEYWORDS

Neurofibromas, hypervascularity

INTRODUCTION: Neurofibromas, pathognomic of neurofibromatosis are complex peripheral nerve sheath tumours. Neurofibromatosis clinically presents with multiple neurofibromas, Lisch nodules in the iris, intertriginous freckling and other ophthalmological manifestations, but hypervascularity is a relatively rare association.

CASE REPORT: A 30 year old female presented with complaints of multiple swellings over the face, trunk and upper limbs for 4 years. The lesions were asymptomatic and there was no positive family history of similar lesions. Dermatological examination revealed multiple firm sessile and pedunculated nodules over the face, trunk and upper limbs. Café au lait macules were appreciated on the trunk and lower limbs. She also had bilateral palmar and axillary freckling. Ophthalmological examination revealed Lisch nodules in the iris. A biopsy was taken from a pedunculated nodule on the chin and was proved to be a neurofibroma.

Ten days after the initial visit, she presented with an intensely bleeding, erythematous, fleshy and proliferative growth over the chin, at the site of the previously biopsied neurofibroma. Suspecting a pyogenic granuloma, a repeat biopsy was taken from the same site and it showed features of neurofibroma with increased vascularity.

DISCUSSION

Neurofibromatosis is an autosomal dominant[1] multisystem disorder predominantly affecting the nervous system, with benign tumours arising from the nerve cells. The defect is thought to lie in mutations of the tumor suppressor gene, neurofibromin 1(NF1)[2].

Neurofibromatosis type 1 is associated with vascular anomalies like arterio-venous malformations, aneurysms and stenosis and is given the name Neurofibromatosis type 1 vasculopathy[3].

Few studies have mentioned an increased vascularity in neurofibroma and attribute the cause to increased angiogenic factors like vascular endothelial growth factor, platelet-derived growth factor and basic fibroblast growth factor[4]. Studies have also shown evidence of recurrent pyogenic granulomas in patients with neurofibromatosis and Von Hippel Lindau syndrome[5]. The common feature in these three conditions is the increase in vascular endothelial growth factor[6].

Histopathology of a neurofibroma will show hypercellular dermis with non encapsulated, well circumscribed dermal tumours consisting of spindle cells with elongated nuclei arranged in eddies and whorls amidst thin collagenous strands. In the presence of an arterio-venous malformation, the dermal vessels will be dilated, proliferative with extravasated RBC's.

Treatment of neurofibromatosis is multidisciplinary and dermatological treatment of the neurofibromas include topical sirolimus and imatinib mesylate[7] and oral agents like ketotifen and pirfenidone[8].

CONCLUSION

Removal of large neurofibromas can result in haemorrhage due to hypervascularity and neurofibromatosis type 1 vasculopathy and this should be borne in mind since there are few studies indicating the same.

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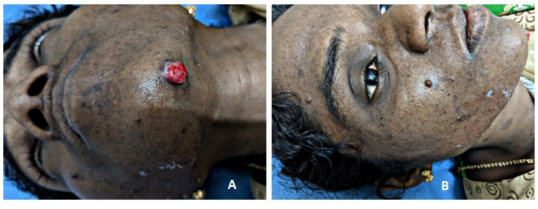

Figure 1: A-Erythematous, fleshy, proliferative growth at the site of the previously biopsied neurofibroma(chin) B-Neurofibroma on the right cheek

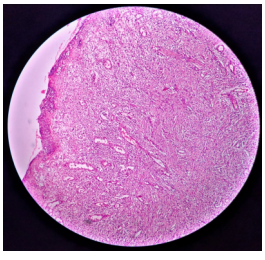

Figure 2: Low power view of the erythematous chin lesion showing epidermal atrophy, hypervascularity and eddies and whorls of spindle cells with wavy nuclei.

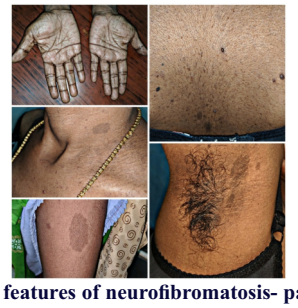

Figure 3: Other features of neurofibromatosis- palmar freckling, café au lait macules, axillary freckling and neurofibromas at other sites.

REFERENCES:

1. McClatchey AI. Neurofibromatosis. Annu. Rev. Pathol. Mech. Dis.. 2007 Feb 28;2:191-216.
2. Viskochil D, Buchberg AM, Xu G, Cawthon RM, Stevens J, Wolff RK, Culver M, Carey JC, Copeland NG, Jenkins NA, White R. Deletions and a translocation interrupt a cloned gene at the neurofibromatosis type 1 locus. Cell. 1990 Jul 13;62(1):187-92.
3. Oderich GS, Sullivan TM, Bower TC, Gloviczki P, Miller DV, Babovic-Vuksanovic D, Macedo TA, Stanson A. Vascular abnormalities in patients with neurofibromatosis syndrome type I: clinical spectrum, management, and results. Journal of vascular surgery. 2007 Sep 1;46(3):475-84.
4. Kawachi Y, Xu X, Ichikawa E, Imakado S, Otsuka F. Expression of angiogenic factors in neurofibromas. Experimental dermatology. 2003 Aug;12(4):412-7.
5. Haitz K, Mikailov A, Foreman R, Zimarowski MJ, Seo SJ. Recurrent lobular capillary hemangiomas in a patient with neurofibromatosis type 1 (NF1) and von Hippel-Lindau syndrome (VHL). JAAD case reports. 2015 Nov 1;1(6):368-70.
6. Lonser RR, Glenn GM, Walther M, Chew EY, Libutti SK, Linehan WM, Oldfield EH. von Hippel-Lindau disease. The Lancet. 2003 Jun 14;361(9374):2059-67.
7. Yang FC, Ingram DA, Chen S, Zhu Y, Yuan J, Li X, Yang X, Knowles S, Horn W, Li Y, Zhang S. Nf1-dependent tumors require a microenvironment containing Nf1+/−-and c-kit-dependent bone marrow. Cell. 2008 Oct 31;135(3):437-48.
8. Widemann BC, Babovic-Vuksanovic D, Dombi E, Wolters PL, Goldman S, Martin S, Goodwin A, Goodspeed W, Kieran MW, Cohen B, Blaney SM. Phase II trial of pirfenidone in children and young adults with neurofibromatosis type 1 and progressive plexiform neurofibromas. Pediatric blood & cancer. 2014 Sep;61(9):1598-602.